

Turner Syndrome and Attention-Deficit with Hyperactivity Disorder in a Pediatric Population

Síndrome de Turner e Perturbação de Défice de Atenção e Hiperatividade numa População Pediátrica

Tânia B Cavaco^{1*}, Marta Novo², Sara Soares³, Inês Vaz Matos⁴, Diana Gonzaga⁵, Leonilde Machado⁶, Teresa Borges⁷, Catarina Prior⁸

*Autor Correspondente/Corresponding Author:

Tânia B Cavaco (tania_cavaco@hotmail.com)

ORCID ID: <https://orcid.org/0000-0002-1553-7244>

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Attention-deficit/ hyperactivity disorder (ADHD) is defined by impairing symptoms of inattention and/or hyperactivity and impulsivity, affecting academic, social, and emotional functioning, often presenting in childhood.^{1,2} This disorder results from the interaction of genetic and environmental factors, and it has been linked to several single gene defects and aneuploidies, including Turner syndrome (TS).³ The diagnosis is based on DSM-5-TR™ criteria² and may be supported by several diagnostic tools, such as Conners Rating Scales (CS).

TS is a genetic disorder resulting from total or partial monosomy of the X chromosome and one of the main causes of ovarian insufficiency and short stature, in females. Besides some typical physical features, these girls also have a distinctive neurocognitive profile

(NCP), characterized by deficits in social cognition, difficulties in problem solving task (e.g., mathematics) and visuospatial organization, with preserved verbal skills. They also have a greater risk of suffering from ADHD and other executive functions disturbances.⁴

A descriptive cross-sectional study was conducted in a III level hospital to better characterize the NCP of TS. This study focused on assessing the presence of ADHD symptoms in pediatric patients with TS, followed up in Endocrinology consultations, according to DSM-5-TR™ criteria and CS' results from parents and teachers.

This study included a sample of 15 patients with TS, with an average age of 13 years and seven months,

1. Child and Adolescent Psychiatry Service, Hospital Garcia da Orta, Almada, Portugal. **2.** Pediatric Department, Centro Hospitalar Universitário do Algarve, Faro, Portugal. **3.** Neurodevelopment Unit, Pediatric Department, Centro Materno Infantil do Norte/ Centro Hospitalar Universitário de Santo António, Porto, Portugal. **4.** Neurodevelopment Unit, Pediatric Department, Centro Materno Infantil do Norte/ Centro Hospitalar Universitário de Santo António, Porto, Portugal. **5.** Neurodevelopment Unit, Pediatric Department, Centro Materno Infantil do Norte/ Centro Hospitalar Universitário de Santo António, Porto, Portugal. **6.** Pediatric Department, Centro Hospitalar Tâmega e Sousa, Penafiel, Portugal. **7.** Pediatric Endocrinology Unit, Pediatric Department, Centro Materno Infantil do Norte/ Centro Hospitalar Universitário de Santo António, Porto, Portugal. **8.** Neurodevelopment Unit, Pediatric Department, Centro Materno Infantil do Norte/ Centro Hospitalar Universitário de Santo António, Porto, Portugal

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who attended from preschool to the 11th grade in school. Regarding school performance, 73% (n=11/15) had never had a grade retention, and 27% (n=4/15, two of which had grade retentions) had educative support measures. Sixty percent (n=9/15) had difficulties in mathematics, 20% (n=3/15) presented difficulty with the Portuguese language, and 13% (n=2/15) had an excellent academic performance. Forty-seven percent attended speech therapy, 40% have had psychology support and 27% attended occupational therapy.

Based on DSM-5-TR™ criteria, ADHD was considered in four patients (26.7%), two of which had an inattention presentation, one had a hyperactivity-impulsivity profile, and one displayed a combined presentation. Regarding CS assessment, 11 parents and seven teachers have collaborated by submitting their observations. According to the parents' consideration, four girls were identified as having ADHD (50% were on P74-85, and 50% on P94-97), while teachers had identified only one case, on P85-89. Three of these four patients were already previously diagnosed with ADHD and under psychostimulants with good therapeutic response. Accordingly, one patient was referred to our medical Neurodevelopmental Consultation following this study.

Despite our reduced sample size, and the scarcity of teachers' CS assessment, the global prevalence of ADHD in our patients' sample with TS is identical (27%) to that described in the literature (24%).⁵ Nevertheless, when considering only results from parents' CS and DSM-5 TR™ criteria, the prevalence may, in fact, be higher (n=4/11, 36%).

The authors aim to highlight the importance of referring patients with TS to a neurodevelopmental consultation, and an annual neurodevelopmental evaluation, even in the absence of obvious complaints. This assessment may uncover subtle neurodevelopmental deficits which, if improved, can significantly optimize these patients' performance, wellbeing, and quality of living.

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TC AND MN: Design, acquisition, analysis, and interpretation of data; Drafting the article; Final approval of the version to be published; Agreement in taking responsibility for the accuracy and integrity of the work.

SS, IM, DG, LM, TB AND CP: Conception and design, analysis, and interpretation of data; Critically revising

the article; Final approval of the version to be published; Agreement in taking responsibility for the accuracy and integrity of the work.

TC E MN: Conceção, aquisição, análise e interpretação dos dados; Redação do artigo; Aprovação final da versão a ser publicada; Concordância em assumir a responsabilidade pela exatidão e integridade do trabalho.

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